Letter to the Editor

Williams Syndrome in Slovakia

To the Editor:

Slovakia is the only postcommunist country in Central Europe with a Williams Syndrome Society. Experience obtained during the 5 years of existence of the Society has clearly demonstrated its usefulness for the affected children and their families [Bzdúch and Sadloňová, 1994].

The concentration of more than 30 individuals suffering from this rare disease has helped enhance clinical research on the disease.

We briefly present some results of this research that have contributed to expanding the knowledge of Williams syndrome:

1. Using two-dimensional echocardiography we established the presence of supravalvular aortic stenosis as a constant finding in Williams syndrome [Bzdúch and Mašura, 1993].
2. We contributed to widening the phenotypic spectrum of Williams syndrome through documentation of radioulnar synostosis [Bzdúch, 1994].
3. We detected a rare case of a child in the hypercalcemic phase of this disease, in whom we established the levels of hormones regulating calcium metabolism [Bzdúch, 1993].
4. We established basic differential diagnostic differences between Noonan and Williams syndromes [Bzdúch, 1995].

Today, Williams syndrome can be diagnosed in Slovakia, as in other developed countries, by DNA analysis (unpublished data).

In conclusion, the establishment of an active Williams Syndrome Society has contributed, with the help of parents and specialists, to the attainment of two objectives: improving care for affected children, and intensifying research on the disease.

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